Genetics Uncoded:

Facts about

LAMB3-Related Junctional Epidermolysis Bullosa

What Your Test Results Mean

Carriers typically show no symptoms of LAMB3-related junctional epidermolysis bullosa (JEB); however, carriers are at an increased risk of having a child with JEB. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

LAMB3-Related Junctional Epidermolysis Bullosa Explained

JEB is an inherited condition in which a subunit of laminin-5 is defective. Laminin-5 is responsible for holding layers of skin together, and when it is non-functional, the skin becomes very fragile and blisters easily. Affected individuals have blistering over large areas of their bodies including the skin, the mouth, and the digestive tract beginning in birth or infancy. Individuals with JEB are very susceptible to infections and may have difficulties with eating and digestion. There is no cure for JEB, and individuals with both the severe Herlitz JEB and more mild non-Herlitz JEB have shortened lifespans.

How the Genetics Work

LAMB3-related JEB is an autosomal recessive skin disorder caused by pathogenic variants in the *LAMB3* gene. In general, individuals have two copies of the *LAMB3* gene. Carriers of JEB have a single variant in one copy of the *LAMB3* gene while individuals with JEB have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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